Fibrinogen Liberec II – dysfibrinogenemia caused by Y211H mutation

E. Ceznerová, J. Štikarová, J. Suttnar, Ž. Sovová, J. Loužil, R. Kotlín, P. Salaj, J. E. Dyr

Introduction: Congenital dysfibrinogenemia is a rare disorder characterized by inherited abnormality resulting in functional disorder of fibrinogen molecule. We describe a new case of fibrinogen mutation in the Czech Republic.

Materials & Methods: A 58-year-old female suffered from a retinal artery occlusion of a right eye. She was diagnosed with heterozygous prothrombin mutation G20210A in 2014. Coagulation tests revealed a decreased level of functional fibrinogen (1.3 g/l), shortened thrombin and reptilase time. DNA sequencing, fibrin polymerization, fibrinolysis, kinetics of fibrinopeptides release and confocal microscopy were performed.

Results: Sanger sequencing of patient’s genomic DNA showed heterozygous point mutation in exon 7 of FGG gene resulting in codon change γ Y211H. Dysfibrinogenemia is presented with normal fibrin polymerization by thrombin and normal fibrinopeptide release. Polymerization curve induced by reptilase had a faster start of clot formation. Confocal laser microscopy revealed thinner fibers, which were significantly different from normal control.

Conclusions: We report a new case of dysfibrinogenemia γ Y211H, which was found by routine coagulation tests and named fibrinogen Liberec II. Mutation γ Y211H has been already described with a combination of Bβ K148R mutation leading to hypofibrinogenemia in fibrinogen Caversham. In our case the mutation has influenced fibrin polymerization.

This work was supported by the project of the Ministry of Health, Czech Republic, for conceptual development of research organization 00023736, by Grant from the Academy of Sciences, Czech Republic nr. P205/12/G118, and by ERDF OPPK CZ.2.16/3.1.00/24001.

Correspondence: Eliška Ceznerová, Institute of Hematology and Blood Transfusion, U Nemocnice 1, 128 20, Praha 2, Czech Republic.
Tel: +420 221 977 345; e-mail: eliska.ceznerova@uhkt.cz

I would like to apply for the Outstanding Abstract Award.